



## GALK1 gene

galactokinase 1

### Normal Function

The *GALK1* gene provides instructions for making an enzyme called galactokinase 1. This enzyme enables the body to process a simple sugar called galactose, which is present in small amounts in many foods. Galactose is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas.

Galactokinase 1 is responsible for one step in a chemical process that converts galactose into other molecules that can be used by the body. Specifically, this enzyme modifies galactose to create a similar molecule called galactose-1-phosphate. A series of additional steps converts galactose-1-phosphate to another simple sugar called glucose, which is the main energy source for most cells. Galactose-1-phosphate can also be converted to a form that is used to build galactose-containing proteins and fats. These modified proteins and fats play critical roles in chemical signaling, building cellular structures, transporting molecules, and producing energy.

### Health Conditions Related to Genetic Changes

#### galactosemia

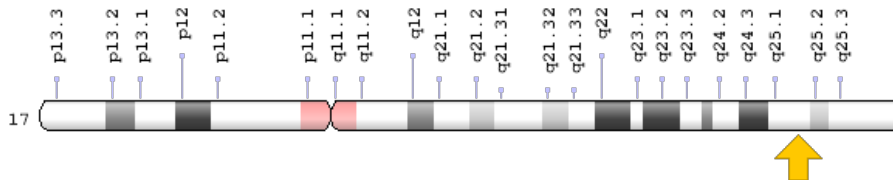
More than 30 mutations in the *GALK1* gene have been identified in people with a form of galactosemia called type II or galactokinase deficiency. Affected infants develop clouding of the lens of the eye (cataracts) but otherwise experience few of the long-term complications associated with classic galactosemia. Most of these mutations change single protein building blocks (amino acids) in galactokinase 1. A few mutations delete a small amount of genetic material from the *GALK1* gene, resulting in an unstable or inactive version of this enzyme.

A shortage of functional galactokinase 1 prevents cells from processing galactose obtained from the diet. As a result, galactose and a related sugar called galactitol can build up, particularly in cells that make up the lens of the eye. An accumulation of these substances damages the lens, causing it to become cloudy and leading to blurred vision.

## Chromosomal Location

Cytogenetic Location: 17q25.1, which is the long (q) arm of chromosome 17 at position 25.1

Molecular Location: base pairs 75,757,937 to 75,765,199 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ATP:D-galactose 1-phosphotransferase
- galactokinase
- GALK
- GALK1\_HUMAN
- GK1

## Additional Information & Resources

### Educational Resources

- Essentials of Glycobiology (second edition, 2009): Figure: UDP-galactose Synthesis and Galactosemia  
<https://www.ncbi.nlm.nih.gov/books/NBK1939/figure/ch42.f3/>

### GeneReviews

- Classic Galactosemia and Clinical Variant Galactosemia  
<https://www.ncbi.nlm.nih.gov/books/NBK1518>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GALK1%5BTIAB%5D%29+OR+%28galactokinase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- GALACTOKINASE 1  
<http://omim.org/entry/604313>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GALK1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4118](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4118)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2584>
- UniProt  
<http://www.uniprot.org/uniprot/P51570>

## **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26143117>

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